PLEASE TAKE THE TIME TO READ AND BE AWARE OF THIS IMPORTANT HEALTH UPDATE IN OUR TREASURED BREED

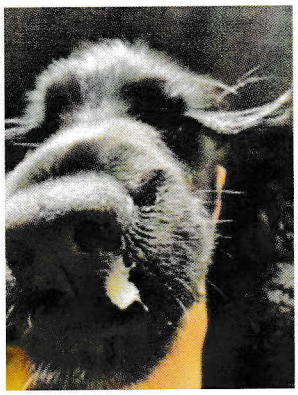
PRIMARY CILIARY DYSKINESIA (PCD)
A HEREDITARY DISEASE DIAGNOSED IN COCKER SPANIELS

The Cocker Spaniel Club of Victoria Inc is committed to improving the quality of the breed, including eliminating any hereditary diseases. Thanks to a bequest we are able to support a genetic study for the development of a DNA test for Primary Ciliary Dyskinesia in Cocker Spaniels.

WHAT IS PRIMARY CILIARY DYSKINESIA?

Dog Primary Ciliary Dyskinesia (PCD) is an autosomal recessive inherited disorder that causes defects in cilia in the upper and lower respiratory tract. In simple terms, in the airway there are tiny hairs (cilia) that are constantly moving mucus. In normal airways, the cilia move the mucus up the respiratory tract to the back of the throat where the mucus with any foreign bodies is then swallowed and sent down the oesophagus into the stomach. In PCD sufferers, those cilia don't function properly, so there is a far greater risk of foreign bodies, such as bacteria or dust ending up in the lungs and pneumonia developing. PCD sufferers can also have situ inversus, which is the condition where internal organs such as the heart are reversed and are in mirror positions to where they would normally be.

Clinical signs in dogs are predominately respiratory in nature, most commonly including chronic nasal discharge and recurrent lower respiratory tract infections commencing in the neonatal period.



An affected pup with a constant flow of chronic discharge

Symptoms of PCD include:

- Chronic mucoid nasal discharge
- Sneezing
- Coughing
- Exercise intolerance
- Respiratory distress
- Cyanosis (blue/purple discolouration of the tongue and gums)
- Deafness and/or loss of balance due to fluid build-up in the middle ear
- Infertility due to altered sperm motility.

Eventually, without the ability to protect their respiratory tract these puppies will suffer from chronic respiratory infections, (including pneumonia) and require ongoing/frequent antibiotic treatment. Adult dogs may have long term sinus infections that don't resolve even with appropriate antibiotic use.

CARE AND MANAGEMENT

Pups that survive experience:

- Daily, life-long nasal discharge and chronic, often wet/productive cough
- Increased risk of acute symptoms due to the development of secondary infections
- Ongoing/frequent antibiotic treatment.

The prognosis for affected pups is guarded to poor. Life-expectancy will be significantly reduced due to the gradual loss of lung function and the potential for resistant/refractory infections (eg life-threatening pneumonia).

Care and management involve:

- Daily chest physiotherapy – coupage
- Daily exercise (which helps to shift mucus)
- Daily nasal saline sprays or daily saline nebulisation which may also help to humidify the airways and mobilise mucus
- Minimum exposure to inhaled irritants/allergens (eg dust, smoke, pollens, perfumes, aerosol sprays, incense) as these particles may get trapped in the mucus and trigger an inflammatory reaction
- Mucolytic medications (designed to reduce mucus build up)
- Periodical thoracic radiographs (every 6-12mths) to monitor for bronchiectasis (dilated airway) and pneumonia
- Bacterial culture of nasal swab +/- bronchoalveolar lavage (BAL, lung wash) samples with any worsening of symptoms

GENETICS

Dog Primary Ciliary Dyskinesia (PCD) is inherited as an autosomal recessive disorder, meaning that the affected pup must receive two copies of the mutated (abnormal) gene, one from each parent, to develop the disease, just like PRA. A dog carrying one copy of the mutated gene is heterozygous and will not show the PCD symptoms. When mating two carriers (heterozygotes) at conception each pup has a 25% chance of being affected, a 50% chance of being an asymptomatic carrier, and a 25% chance of being unaffected and not a carrier. Currently there is no cure for PCD and the only way to avoid breeding affected pups is to breed dogs which are not carriers of the mutation. Defective genes for autosomal recessive disease can be passed for many generations without affected individuals occurring until two carriers are bred to one another. The only way to find out if there is a chance of getting an affected puppy is to do genetic testing. **Currently there is no DNA test available for PCD in Cocker Spaniels.**

INCIDENCE AND FACTS OF PCD IN COCKER SPANIELS IN AUSTRALIA

PCD has been identified in more than 19 breeds, including Cocker Spaniels. Cases of PCD in Cocker Spaniels have been officially recorded in Australia from 2013 in different States and over different generations.

Two cases of Cocker Spaniels with PCD were published in the Australian Veterinary Journal in an article written by E Bell in 2016. One of the dogs mentioned in that study is now 8 and undergoes an extensive management regime to maintain her health and wellbeing. The other pup diagnosed was euthanised due to the severity of the disease.

Other Cocker Spaniels have also been diagnosed since that report with 3 pups being diagnosed in 2022.

It is likely that PCD is more common in Cocker Spaniels than reported, because affected dogs may be misdiagnosed as having fading puppy syndrome, aspiration pneumonia, or infectious causes of pneumonia or they may be euthanised at a young age due to chronic respiratory infections without being diagnosed.

There are several other reported suspected cases of PCD in young Cocker Spaniels who presented with recurrent respiratory signs, however official diagnosis has not been carried out due to the expense and difficulty in diagnosis.

RESEARCH AND GENETIC TRIALING

Currently, there is no reasonable way to distinguish carriers from normal dogs because they look the same and do not have the disease. A DNA-based test would enable the identification of carriers so as not to breed a carrier to carrier and eliminate PCD from future breeding generations.

A genetic study into the development of a DNA test for PCD in Cocker Spaniels is being conducted by a project team through the University of Sydney and overseen by Professor Claire Wade BSc (Hons) PhD UNSW GCertEd UQ. Chair of Computational Biology and Animal Genetics Sub Dean International in consultation with Dr Lydia Hambrook BVSc (Hons), FANZCVS (Registered Specialist in Small Animal Medicine).

The purpose of this information is to raise awareness of the existence of PCD in the breed to our sister clubs, breeders and members and to invite those who may wish to do so, to participate in the trial.

For further information on how to participate please see the contact details below:

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Participating in the trial is a simple as having a blood sample taken. All information pertaining to dogs, blood samples and pedigrees is totally CONFIDENTIAL and remains so with regard to any information that may be published in the findings of the trial.

With the development of a DNA test a simple swab sample will determine the status of dogs as being clear or carrier and hence enable breeders to eliminate this horrific disease from the breed as has been achieved with other known inherited diseases.

For further information about PCD you can also contact our PCD Liaison Officer
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COCKER SPANIEL CLUB OF VIC. (INC.)